Neuroacanthocytosis

National Institute of Neurological Disorders and Stroke (NINDS)

Source

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Neuroacanthocytosis refers to a group of genetic conditions that are characterized by movement disorders and acanthocytosis (abnormal, spiculated red blood cells). Four syndromes are classified as neuroacanthocytosis: Chorea-acanthocytosis, McLeod syndrome, Huntington's disease-like 2 (HDL2), and panthothenate kinase-associated neurodegeneration (PKAN). Acanthocytosis may not always be observed in HDL2 and PKAN. These disorders are caused by different genetic mutations, and the signs and symptoms vary, but usually include chorea (involuntary, dance-like movements), parkinsonism (slowness of movement), dystonia (abnormal body postures), and problems walking. There may also be muscle weakness, involuntary movements of the face and tongue, tongue/lip biting (which is mostly characteristic of Chorea-acanthocytosis), as well as difficulty with speech and eating, cognitive impairment, psychiatric symptoms, and seizures. Individuals with McLeod syndrome often have cardiac problems. Many features of these disorders are due to degeneration of the basal ganglia, a part of the brain that controls movement. Additional disorders that are also known have neurologic symptoms, acanthocytosis, and either lipoprotein disorders or systemic findings. The diagnosis of neuroacanthocytosis is typically based on the symptoms and clinical observation, a review of family history, and the evaluation of specific laboratory and imaging studies.